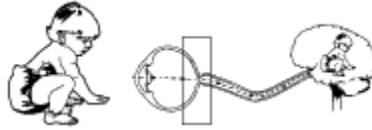


## ALBINISM



## DEFINITION

Albinism refers to a group of inherited conditions. People with albinism have absent or reduced pigment in their eyes, skin or hair. They have inherited genes that do not make the usual amounts of a pigment called melanin which is essential for the full development of the retina. Lack of melanin in development of the retina is the primary cause of visual impairment in albinism. In the USA it is estimated that one person in 17,000 has some type of albinism.

There are two major categories of albinism (overlap of these categories often occurs):

**Ocular Albinism (OA)** is divided into two types according to the inheritance pattern:

**autosomal recessive OA** occurring equally in males and females, and **X-linked OA** with symptoms occurring primarily in males. In the X-linked cases, mothers carry the gene and pass it to their sons. Although the mothers usually have normal vision, they have subtle eye changes that can be identified by an ophthalmologist. If a woman does carry the gene, with each pregnancy there is a one in two chance of having a son with X-linked ocular albinism. For specific information, families should seek the advice of a qualified genetic counselor.

**Oculocutaneous Albinism (OCA)**, involves the lack of pigment in the hair, skin and eyes. Each parent must carry the gene for this form which follows an autosomal recessive inheritance pattern, meaning there is a one in four chance at each pregnancy that the baby will be born with albinism. Within OCA there are two primary types of albinism.

Type 1 (formerly known as Tyrosinase Negative) involves the complete lack of pigment. These children have white skin and hair and moderate to severe visual impairment. Children with Type 2 (formerly known as Tyrosinase Positive) have various amounts of pigmentation, yellow or blonde hair and usually less severe visual impairment.

## **Secondary OCA**

The appearance of albinism may indicate the presence of other rare conditions that require special management. Hermansky-Pudlak Syndrome (HPS) involves a platelet defect that results in susceptibility to bleeding and bruising. Aspirin and aspirin-like drugs should be avoided, as they may convert a mild bleeding disorder into a severe one. HPS can be identified by a specialized test involving electron microscopy of the platelets. Chediak-Higashi Syndrome is associated with a white blood cell problem leading to a susceptibility to bacterial infections. Prior to a surgical procedure for persons with these rare forms of albinism, specific tests for bleeding dysfunction and white blood cell abnormalities should be performed.

## **DIAGNOSIS**

The diagnosis of albinism can be challenging. If albinism is suspected, concerns should be communicated to the pediatrician who will refer the family to a pediatric ophthalmologist. If a child has OCA, diagnosis will often be based first on the fair color of the child's skin and hair. A diagnosis of albinism may be suspected when a child develops rhythmic, involuntary eye movements (nystagmus) within the first few weeks of life and the eye exam identifies:

- if light passes through the iris (transillumination)
- the underdevelopment of the central retina (foveal hypoplasia), and
- the absence of melanin pigment in the eye.

While hair bulb testing has been used as a diagnostic test for albinism in the past, newer, more accurate genetic tests are now available for diagnosis, classification, and prenatal genetic counseling. The visual evoked potential test (VEP), designed to identify the misrouting of retinal fibers of the optic nerve, can be valuable in providing a specific diagnosis.

## **CHARACTERISTICS**

- Low Vision (visual acuity between 20/50 and 20/800)
- Sensitivity to bright light and glare (photophobia)
- Rhythmic, involuntary eye movements (nystagmus)
- Absent or decreased pigment in the skin and eye and sensitivity to sunburn (ultraviolet light) that could lead to skin cancers or cataracts in later life
- "Slowness to see" in infancy
- Farsighted, nearsighted, often with astigmatism
- Underdevelopment of the central retina (foveal hypoplasia)
- Decreased pigment in the retina (blonde fundus)
- Inability of the eyes to work together (absence of stereo vision)
- Misrouting of the nerve pathways from the retina to the brain
- Light colored eyes ranging from lavender to hazel, with the majority being blue
- Strabismus, with both vertical and horizontal deviations

## VISUAL AND BEHAVIORAL CHARACTERISTICS

- Infants with albinism may behave as if they are not seeing during the first weeks of life and gradually become visually attentive. This is now a well-documented condition.
- Additional energy and effort a child uses to process visual information can cause fatigue and irritability, worsening nystagmus.
- Children typically exhibit an eye and/or head position that allows them to slow down or stop their nystagmus (null point).
- Children may use one eye at a time for looking.
- They may have an absence of stereo vision contributing to depth perception problems.
- Children bring objects close to their eyes to see more clearly.
- Even with glasses or contacts, vision will not correct to normal.
- Occasionally a child's eyes will look pink or red due to a reflection on the back of the eye (retina).
- The following environmental factors can have a significant influence on the child's ability to see: changes in lighting from various light sources, the position of the light source, and glare from reflecting surfaces. Looking directly into a bright light or window creates a silhouetting effect, diminishing the ability to see detail. Children are also slow to adjust to changes in lighting, such as from outdoors to indoors or sunny light to shadows.

## MYTHS

The following statements are **NOT TRUE**, according to current knowledge in the field:

- Persons with albinism always have red eyes.
- Persons with albinism are totally blind.
- Albinism is contagious.
- Persons with albinism are the result of evil spirits or wrongdoing.
- Persons with albinism are retarded or deaf.
- Albinism results from inbreeding or the mixture of two races.
- Persons with albinism have magical powers.

## SOCIAL AND EMOTIONAL ISSUES

Persons with albinism often have quite normal lives. The support of family, teachers, and friends is crucial in order for a child with albinism to grow up with a positive self image. Sometimes, however, the appearance of the person with albinism becomes a focus, rather than the individual. A person with albinism may try to minimize the differences caused by albinism, resulting in a great deal of stress at continually trying to maximize visual ability and/or denying altogether that one even has albinism. Families of children with albinism may experience prejudice, rejection, or frustration. They may become caught in isolation and denial. As well, the myths related to albinism can interfere with the family's free discussion of the condition.

## TEACHING STRATEGIES

- Since not all children with albinism have the same visual acuity or use their vision with the same efficiency, it is important to consider each child as an individual.
- Early and ongoing assessment of the child's vision is crucial. As children progress through the educational system, they face increased visual demands and smaller print size and thus can benefit from suitable magnifiers, telescopes, or large print materials.
- It is important that the child, parent, teacher, vision resource teacher, ophthalmologist, and optometrist work as a team. The team should consider positioning, seating, lighting, glare, toys and materials, along with social/emotional growth of each child.
- The condition of "slowness to see" calls for an approach that uses all the senses for learning (multisensory) with the very young child.
- Allow the child to use the head and eye position that is comfortable and works the best.
- Provide good indirect lighting or position a light source behind the child. Never position a child directly facing the light source. Also, parents or teachers should always position themselves so that the child is not looking toward a light source.
- A child who may be poorly organized may benefit from predictable environments and routines.
- A child should be able to hold toys and objects as close as he would like and should be allowed to move to the most advantageous viewing position.
- A careful "walk-through" of the house or classroom will identify the areas of highest glare. Using indoor lighting, even during the day, can equalize lighting from indoor and outdoor sources and reduce the effects of glare on the eyes. A hat or visor can eliminate glare from overhead lighting.
- To compensate for missed nonverbal cues such as facial expression, and everyday social gestures, etc., a child should be given verbal and/or tactual information.
- Lack of depth perception can be compensated for by learning to use information from the environment. Provide a child with repeated opportunities for exploration and movement in a variety of settings and lighting conditions.
- When the environment makes it difficult to see, a child must learn to rely on other senses. Bright glare from snow, sand, water, or pavement, or dappled light where sun and shadows continually change, can reduce a child's visual world.
- Open discussion and support at home for a child with albinism encourages him to acknowledge his feelings and to begin the process of self-advocacy.
- The following qualities make a significant difference in the child's ability to see printed materials:
  - High contrast Uncluttered background
  - Well defined pictures
  - Large print Primary colors

## GLOSSARY

1. **Electron Microscopy:** examination by a microscope which uses a beam of electrons to form an enlarged image of a specimen.
2. **Platelets:** one of three types of blood cells. The platelet is the smallest cellular element of the blood and is needed for proper clotting.
3. **Refractive Error:** an eye defect that causes decreased visual acuity.
4. **Stereo Vision:** the perception of objects in space and their relative position to one another.

## RESOURCES

Haefemeyer, J. (1997). Visual Problems in Albinism, NOAH News, Winter.

Haefemeyer, J. (1996). X-Linked Ocular Albinism Researched, NOAH News, Winter.

Haefemeyer, J., Kind, R., LeRoy, B. (1992). [Facts About Albinism](#)

Johnson Printing Company (1996). Transactions of the American Ophthalmological Society Annual Meeting -1996.

King, R. A., Summers CG. (1988). Albinism, Dermatologic Clinics, 6,217-227.

Kinnear, P., Barrie, J., Witkop, C. (1985). Albinism, Survey of Ophthalmology, 30 (2),75-101.

Waugh, J. (1990). Social and Emotional Aspects of Albinism, NOAH News, Spring.

Moore, Dennis (1991). Another View: Albino vs. Person With Albinism, NOAH News, Spring.

National Organization for Albinism and Hypopigmentation (1992). Assisting Students with Albinism, NOAH pamphlet.

[National Organization for Albinism and Hypopigmentation](#) (1992). What is Albinism, NOAH pamphlet. NOAH, 1530 Locust St. #29, Philadelphia, PA 19102, 1-800-473-2310

## **ACKNOWLEDGMENTS**

Project Coordinator: Julie Bernas-Pierce, M.Ed.  
Dr. Doug Fredrick, Nancy Akeson, Susan Gomez, June Waugh,  
Michele Daly, Hsiao-hui Ning, Gail Calvello.  
Reviewers: Dr. Jim Haefemeyer, Kathryn Neale Manalo

The Pediatric Visual Diagnosis Fact Sheets are sponsored by a grant from the Blind Children's Center and with support from the Hilton/Perkins Program through a grant from the Conrad Hilton Foundation of Reno, Nevada.

REPRODUCTION FOR RESALE IS STRICTLY PROHIBITED (1/98 BBF)



### **BLIND BABIES FOUNDATION**

1814 Franklin St, 11<sup>th</sup> Floor  
Oakland, CA 94612 (510)446-2229  
[www.blindbabies.org](http://www.blindbabies.org)